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Editorial Comment

Microcephaly, Muscular Build, Rhizomelia and Cataracts: A Possible Recessive Syndrome

The analysis of performance of LDDB and POSSUM by Verloes et al. is of considerable value and points up some of the pitfalls of using such databases. The authors begin by searching under single traits. Of course, one would never do this as part of a normal search as too many syndromes are found. The authors then do a combinatorial search on 8–9 manifestations and get a reasonably useful differential diagnostic list. They query why Smith-Lemli-Optiz syndrome type II was not retrieved by LDDB. This is because we do not have this syndrome coded under "micropenis"; it is coded under hypospadias and ambiguous genitalia. This is obviously a question of definition of clinical signs, which is one of the major problems with such databases.

A further way of searching is to pick out two or three rare key anomalies and to do a search on these. The authors suggest that one combination of defects to delineate the present syndrome is an advanced bone age and a muscular build. Searching under these two aberrations retrieves 3 syndromes, none of which seem reasonable diagnostic suggestions. However, one might approach this case differently thinking that the most useful descriptors are cataracts and 2–3 toe syndactyly (the latter is common in a mild form but in the present case there is a substantial degree of syndactyly). Searching on this combination retrieves 12 syndromes, including a case report by Pavone et al. [1993], which was not retrieved by Verloes et al. and which is a closer fit than many of the syndromes mentioned in the paper. Adding hypotelorism to the search, together with syndactyly 2–3 of toes, retrieves the case reported by Harrod et al. [1977], which, again, is a useful suggestion in the differential diagnosis but was not retrieved by Verloes et al. Reference to these papers might provide further insights into the differential diagnosis.

The conclusion to all this is that there are several different strategies for searching such databases and if one does not make a precise diagnosis with one combination of findings, different combinations should be tried to see if the differential diagnosis list can be improved. Experience certainly helps in using these databases and there is a definite "learning curve."

As I have stressed before, the databases are "systems for experts" rather than "expert systems."

REFERENCES

Harrod MJ, Keele DK, Howard J Sr (1977): A syndrome of craniofacial, digital and genital anomalies. BDOAS 13(3B):111–115.

Pavone L, Fiumara A, Rizzo R et al. (1993): Syndactyly type 1 with cataracts and mental retardation. Clin Dysmorphol 2:257–259.

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